



TECPR2 gene

tectonin beta-propeller repeat containing 2

Normal Function

The *TECPR2* gene provides instructions for making a protein that is involved in a cellular process called autophagy. Cells use this process to recycle worn-out or unnecessary cell parts and break down certain proteins when they are no longer needed. During autophagy, materials that are no longer needed are isolated in compartments called autophagosomes. The autophagosomes are then transported to cell structures that break the materials down. The TECPR2 protein is thought to be important for the formation of autophagosomes.

Health Conditions Related to Genetic Changes

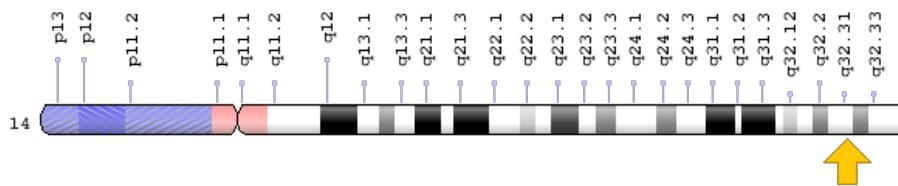
Spastic paraplegia type 49

At least five mutations in the *TECPR2* gene have been found to cause spastic paraplegia type 49. This condition is characterized by gradually worsening muscle stiffness (spasticity) and paralysis in the lower limbs (paraplegia), intellectual disability, and other neurological problems that lead to trouble regulating involuntary body processes, such as digestion and breathing. Many of the mutations that cause spastic paraplegia type 49 change single protein building blocks (amino acids) in the TECPR2 protein. Others lead to production of an abnormally short protein that is likely broken down quickly. Alteration or loss of the TECPR2 protein is thought to impair autophagy, making cells less efficient at removing unneeded materials. Researchers suggest that nerve cells (neurons) may be particularly vulnerable to impaired autophagy because it is especially difficult to transport waste materials through their long extensions (axons and dendrites) for breakdown. The waste materials can build up in neurons and damage them. Damage to neurons results in the neurological problems that occur in spastic paraplegia type 49.

Chromosomal Location

Cytogenetic Location: 14q32.31, which is the long (q) arm of chromosome 14 at position 32.31

Molecular Location: base pairs 102,362,941 to 102,502,477 on chromosome 14 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIAA0329
- SPG49
- tectonin beta-propeller repeat-containing protein 2 isoform 1
- tectonin beta-propeller repeat-containing protein 2 isoform 2

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database (2000): Macroautophagy in Mammalian Cells
<https://www.ncbi.nlm.nih.gov/books/NBK6211/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TECPR2%5BTIAB%5D%29+OR+%28tectonin+beta-propeller+repeat+containing+2%5BTIAB%5D%29+OR+%28tectonin+beta-propeller+repeat-containing+protein+2+isoform+1%5BTIAB%5D%29+OR+%28tectonin+beta-propeller+repeat-containing+protein+2+isoform+2%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- TECTONIN BETA-PROPELLER REPEAT-CONTAINING PROTEIN 2
<http://omim.org/entry/615000>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TECPR2%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:19957
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:9895>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9895>
- UniProt
<https://www.uniprot.org/uniprot/O15040>

Sources for This Summary

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- OMIM: TECTONIN BETA-PROPELLER REPEAT-CONTAINING PROTEIN 2
<http://omim.org/entry/615000>

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